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Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	[redacted]
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Cystic Fibrosis (CFTR) Sequencing with Reflex to Deletion/Duplication

ARUP test code 0051640

Cystic Fibrosis Seq, w/Rflx DelDup Spec	Whole Blood
Cystic Fibrosis Seq, w/Rflx DelDup Int	Negative

TEST PERFORMED - 0051640
TEST DESCRIPTION - Cystic Fibrosis (CFTR) Sequencing with Reflex to Deletion/Duplication
INDICATION FOR TEST - Not Provided

RESULT
No pathogenic variants were detected in the CFTR gene.

INTERPRETATION
No pathogenic CFTR gene variants were detected using bidirectional sequencing of the coding regions and intron-exon boundaries or deletion/duplication analysis. Although this result does not exclude cystic fibrosis (CF) disease or carrier status, the risk for such is greatly reduced.

RECOMMENDATIONS
Medical management should rely on clinical findings and family history. Genetic consultation is recommended.

COMMENTS
Reference Sequence: GenBank # NM_000492.3 (CFTR)
Nucleotide numbering begins at the "A" of the ATG initiation codon.
Likely benign and benign variants are not included in this report.

This result has been reviewed and approved by Wei Shen, Ph.D.

H=High, L=Low, *=Abnormal, C=Critical

BACKGROUND INFORMATION: Cystic Fibrosis (CFTR) Sequencing with Reflex to Deletion/Duplication
CHARACTERISTICS: Chronic sino-pulmonary disease, gastrointestinal malabsorption/pancreatic insufficiency, and obstructive azoospermia. Findings are often limited to a single organ system such as isolated pancreatitis, bilateral absence of the vas deferens, nasal polyposis, or bronchiectasis in non-classic cystic fibrosis (CF).
INCIDENCE OF CLASSIC CF: 1 in 3,000 Caucasians or Ashkenazi Jewish, 1 in 8,000 Hispanics, 1 in 15,000 African Americans, 1 in 32,000 Asians.
INCIDENCE OF NONCLASSIC CF: Unknown.
INHERITANCE: Autosomal recessive.
PENETRANCE: High for severe mutations, variable for mild/moderate mutations.
CAUSE OF CLASSIC CF: Two deleterious CFTR mutations on opposite chromosomes.
CAUSE OF NONCLASSIC CF: Typically one severe and one mild/moderate CFTR mutations on opposite chromosomes.
MUTATIONS TESTED: Base pair substitutions and deletions/duplications within the coding region and intron-exon boundaries; additionally, two deep intronic mutations (3849+10kbC>T and 1811+1.6kbA>G).
CLINICAL SENSITIVITY: 99 percent.
METHODOLOGY FOR SEQUENCING: Bidirectional sequencing of the entire CFTR coding region, intron-exon boundaries and two deep intronic mutations.
METHODOLOGY FOR DELETION/DUPLICATION: Multiplex ligation-dependent probe amplification (MLPA) to detect large CFTR coding region deletions/duplications.
ANALYTICAL SENSITIVITY AND SPECIFICITY FOR SEQUENCING: 99 percent.
ANALYTICAL SENSITIVITY AND SPECIFICITY FOR MLPA: 98 percent.
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Breakpoints for large deletions/duplications will not be determined. Regulatory region and some deep intronic mutations will not be detected.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online at www.aruplab.com.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Cystic Fibrosis Seq, w/Rflx DelDup Spec	19-241-141917	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Cystic Fibrosis Seq, w/Rflx DelDup Int	19-241-141917	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical